Tay-Sachs Disease

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A foundation for TSD Research: Harry Hoffman Foundation – Tay Sachs Disease is always fatal in children.

**Tay Sachs Disease**

A baby is born – perfectly healthy. However, you begin to notice that she is not progressing but rather going backwards in development. You notice an increased startle reaction to noise and a “cherry-red spot” in her eyes. She never learns to walk, she never learns to talk, and she eventually reaches a point where she is able to do nothing on her own. She is unable to push herself up or perform basic motor activities; she experiences respiratory problems as entropy progresses quickly. Trapped inside her own mind, her mental and physical abilities slowly deteriorate until she dies at the young age of 4 or 5. She becomes unable to see, unable to swallow, unable to hear. She becomes prone to seizures and dementia, muscle atrophy and paralysis. This baby girl is lacking a single base pair, missing one gene. The lack or mutation of this important gene (that is responsible for the production of the Hexosaminidase A, or “Hex-A” protein) destroyed this child’s chance of having a life. This genetic disease took her life - way before her time.

Tay Sachs disease is a rare genetic disorder that is usually noticed at a very young age, though there are few cases of adult Tay Sachs that are not as life-threatening. The disease is caused by a mutation of one of the child’s proteins causing them to be unable to produce the Hex-A protein (made up of 2 components – the alpha and beta), which breaks down the fatty acids in the brain (GM2 ganglioside). As shown in figure 1.1, the gene for the production of this protein is on chromosome 15. The alpha part of the Hex-A can be a mutated slightly- possibly even a single base mutation (though there are over 90 possible mutations that could cause the disorder) so that it’s overall effectiveness is down to 50%, thus causing the fat accumulation. The base sequence of a gene changes and causes irrevocable damage to the protein; for example, the wrong amino acid could be added to the “polypeptide chain”. These fat cells accumulate immensely in the nerve cells so the children without this protein have no means of keeping this pressure off of their brain. They have excessive deposits of fat acids in their brain that build up over time, only to eventually destroy the brain from the inside out.

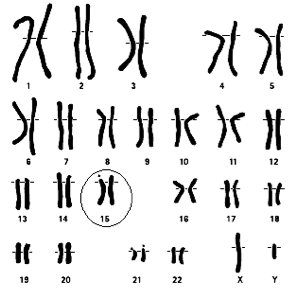
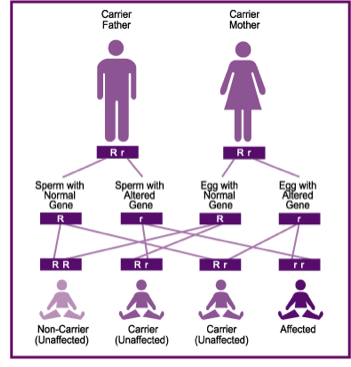


Figure 1.1 – the mutated chromosome 15

Though this disease is relatively rare, the incidence is high and the effects are devastating to families. It is important to be aware of this small mutation with drastic affects – you never think it will happen to you. Tay Sachs is particularly common in individuals of Eastern Europeans and Ashkenazi Jews (about 1 in every 27 Jews is a carrier). Though the disease is also found in other racial groups, in these particular groups, the frequency of the disease is 100 times more than the general population. Compared to an average person’s 1/300 chance of carrying the disease, an Ashkenazi Jew has a 1/30 chance. To avoid this possibility, many opt to take a blood test that measures Beta-Hexosaminidase-A activity – this can allow and individual to understand the risks if they are a carrier. If even one parent is a carrier, the child has a 50% chance of being a carrier - if both parents are carriers, the child has a 25% of contracting the disease. For a child to be affected, she has to receive the mutated gene from both parents. Figure 1.2 breaks down the probabilities of passing on this genetic disease. It is also possible to indentify this disease before birth by testing fluid around the fetus through chorionic villus sampling (CVS). Looking for Hex-A, if the chemical is present, the child will not be affected, however, if it is lacking, the child will have Tay Sachs syndrome. There is no cure or treatment for this disease, no way to stop it from “running it’s fatal course”. There is, however, ways to make the child more comfortable. Anticonvulsant medicine can be used to control seizures while proper nutrition and hydration, along with ways to keep the airway open will be used to save the child for as long as possible. The National Institute of Neurological Disorders and Stroke, or NINDS (part of the National Institutes of Health) funds and conducts extensive research about Tay Sachs and possible preventions and cures. According to the National Health Institute, countless studies are underway testing drugs to break up the fat acids or re-insert the missing proteins. They are currently testing drugs such as Pyrimethamine, Leucovorin, Cyclophosphamide, or even trying to master stem cell transplants.

Figure 1.2 – The probabilities of contraction are illustrated in this diagram.



Maybe the future will bring an end to Tay Sachs. Maybe someday, this horrible disease will meet it’s end and these babies will finally have a chance to live life and live it to the fullest.

Works Cited

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